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Brief Clinical Report

The Floating Harbor Syndrome With Cardiac Septal Defect

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The Floating Harbor syndrome of short stature, very delayed bone age, expressive language delay, and characteristic facial changes has not been associated with cardiac anomalies, except for one patient with pulmonic stenosis. We report on a 10-year-old boy with the syndrome and tetralogy of Fallot with atrial septal defect.

KEY WORDS: Floating Harbor syndrome, cardiac septal defect, pseudoarthrosis of the clavicle, speech and developmental delay

INTRODUCTION

The Floating Harbor syndrome comprises short stature with greatly delayed bone age, expressive language delay usually with normal motor development, and a characteristic triangular face with prominent nose and deep-set eyes. Despite multisystem involvement, cardiac defects have not been documented to occur, except in one case with mild pulmonic stenosis [Patton et al., 1991].

CLINICAL REPORT

The patient is a 10 4/12-year-old white boy, whose healthy 41-year-old mother and a 46-year-old father were of German ancestry without identifiable consanguinity. The patient's older brother had congenital diaphragmatic hernia and was doing well at age 14 years. A younger brother, delivered by emergency cesarean section at 36 weeks due to placental abruptio, had no congenital anomalies. No other relative had birth defects.

The pregnancy was complicated by a low lying placenta, spotting during the second and third trimesters, and small-for-gestational age size. After cesarean section for breech presentation the term infant cried immediately and did not require oxygen. At birth, length was 48 cm (5th centile; 50th centile for 32 gestational weeks), head circumference (OFC) was 31 cm, and weight was 1,720 g (both <5th centile; 50th centile for 32 gestational weeks).

On day 2, a heart murmur was noted. Chest radiograph, electrocardiogram, and echocardiography findings were consistent with tetralogy of Fallot and atrial septal defect. He fed well by bottle without cyanotic spells; blood gas levels were normal in room air. At 2½ months he had bilateral inguinal herniorrhaphies. Catheterization at 3½ years confirmed the diagnosis of tetralogy of Fallot, and total correction was performed without complications.

At 5 years, surgical recession of the medial rectus muscles was done in both eyes to correct esotropia.

Speech and language screening at 3 years 4 months showed delayed speech and language skills. Auditory comprehension was at 2.5 years, and language skills at 12 months. Audiologic tests indicated normal hearing. Toddler and preschool developmental assessment at the age of 3½ years showed overall reasoning and cognitive abilities of a 22- to 23-month-old child.

Skeletal study at 3 years was consistent with a bone age of 6 months, more than 2 standard deviations below the mean. No malformations were seen, but there was an apparent pseudoarthrosis of the right clavicle (Fig. 1). The patient's mother could not recall an episode that may have caused a clavicular fracture. Chromosomes were apparently normal (46,XY at 400 bands). Fragile X was not seen cytogenetically. Thyroid functions were normal.

On physical examination at 10 4/12 years, the patient was a small, thin white male with no cyanosis. Height, weight, and head circumference (and age of 50th centile) were, respectively, 119 cm (6 10/12 years), 18.3 kg (5 2/12 years), and 48 cm (11 months), indicating relative and absolute microcephaly. The outer canthal distance, 7.6 cm (3 6/12), the inner canthal distance, 2.4 cm (6/12), and the interpupillary distance, 4.7 cm (1 2/12), were at

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Fig. 1. Chest radiograph at 3½ years showing hypoplasia of the right clavicle.

the 50th centile for age 3⁶/₁₂, ⁶/₁₂, and 1⁸/₁₂, respectively, and appropriate for head size. The face was triangular, small, and narrow with low posteriorly angulated ears lacking the lobule. He had a short upper lip, thin vermilion border of the lip, micrognathia, very long eyelashes, and a slightly flattened nasal bridge with wide columella. The chest was small but normal. The right clavicle could not be palpated. A grade 3/6 high pitched systolic ejection murmur was best heard at the upper left sternal border. The scapulae were prominent. There was no organomegaly; the umbilicus was not prominent. Genitalia were normal except for well-healed bilateral inguinal scars. Pigmentation and hair distribution were normal for age. The limbs were short in proportional to body size. There was brachydactyly of the 5th digit bilaterally, normal fingernails, and no polydactyly or interdigital webbing.

DISCUSSION

Pelletier and Feingold [1973] and Leisti et al. [1974] each reported a patient with the Floating Harbor syndrome, and with the present cases, the total is 17 [Robinson et al., 1988; Patton et al., 1991; Chudley and Moroz, 1991; Houlston et al., 1994; Majewski and Lenard, 1991] (Table I).

The key to diagnosis (despite prior effort) in this case was the radiographic finding of "pseudoarthrosis" of the right clavicle, in the absence of a history of clavicular fracture. P.O.S.S.U.M. (1994) yielded six syndromes with pseudoarthrosis of the clavicle; three with abnormal chromosomes were excluded. Two other syndromes were Carey-Fineman-Ziter syndrome and mesomelic dysplasia, Reardon-Hall type. The later has many traits not found in our patient. The former syndrome shares many common abnormal traits with the Floating Harbor syndrome. However, it also involves hypotonia, muscle weakness, dysphagia, abnormal neck, and scoliosis, none of which were seen in our patient. Thus, the diagnosis of the Floating Harbor syndrome was established.

The only prior patient with a congenital heart defect had mild pulmonary stenosis [Patton et al., 1991]. Since our patient's more severe cardiac defect likewise reflects underdevelopment of the pulmonary infundibulum, the association may have a biologic basis. Jones and Waldman [1985] reported on a family in

TABLE I. Summary of Abnormalities Reported in Patients With the Floating Harbor Syndrome

	Robinson et al. 1988	Patton et al. 1991	Chudley et al. 1991	Houlston et al. 1993	Majewski et al. 1991	Lazebnik et al. 1995	Total
Male/female	2/4	2/4	0/1	0/2	0/1	1/0	5/12
Growth							
Birth weight < 5%	4/6	1/5	1/1	0/2	?	1/1	7/16
Birth length < 5%	5/6	?	1/1	1/2	1/1	1/1	9/11
Short stature < 2 SD	6/6	?	1/1	2/2	1/1	1/1	11/11
Development							
Delayed motor skills	1/6	2/6	1/1	1/2	0/1	0/1	5/17
Speech delay	6/6	6/6	1/1	2/2	1/1	1/1	17/17
Mental retardation	3/5	1/6	1/1	2/2	?	1/1	8/16
Craniofacial traits							
Bulbous nose	6/6	6/6	0/1	2/2	1/1	1/1	16/17
Wide columella	6/6	6/6	1/1	2/2	1/1	1/1	17/17
Short upper lip	6/6	2/6	1/1	1/2	0/1	1/1	11/17
Thin lips	6/6	4/6	1/1	2/2	1/1	1/1	15/17
Broad thumbs	6/6	3/4	0/1	1/2	1/1	1/1	12/17
Deep set eyes	6/6	1/4	1/1	2/2	0/1	0/1	10/17
Triangular face	2/6	2/6	1/1	1/2	0/1	1/1	7/17
Posteriorly rotated ears	4/6	?	1/1	2/2	0/1	1/1	8/11
Long eyelashes	4/6	?	0/1	2/2	1/1	1/1	7/11
Other findings							
Clinodactyly	4/6	?	1/1	1/2	1/1	1/1	8/11
Brachydactyly	4/6	?	0/1	2/2	1/1	1/1	7/11
Clubbing	2/6	2/6	1/1	1/2	0/1	1/1	7/17
Abdominal distension	4/6	2/6	1/1	?	0/1	0/1	17/15
Coeliac disease	1/6	?	1/1	1/2	?	0/1	3/16
Cardiovascular abnormal	?	1/6	?	?	?	1/1	2/7

which six persons in three successive generations had some combination of preauricular pits (4/6), tetralogy of Fallot (3/6), fifth finger clinodactyly (6/6), and seemingly characteristic facial appearance (5/6) with broad forehead and "prominent" eyes. The absence of a family history of congenital cardiac anomalies and the facial appearance different from our case make it highly unlikely that our patient has the same disorder [Jones and Waldman, 1995].

Thus, pulmonic valvular defects may be a component manifestation of the Floating Harbor syndrome. Suspected cases should have echocardiography.

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